



## KRT12 gene

keratin 12

### Normal Function

The *KRT12* gene provides instructions for making a protein called keratin 12. Keratins are a group of tough, fibrous proteins that form the structural framework of epithelial cells, which are cells that line the surfaces and cavities of the body. Keratin 12 is produced in a tissue on the surface of the eye called the corneal epithelium. This tissue forms the outermost layer of the cornea, which is the clear front covering of the eye. The corneal epithelium acts as a barrier to help prevent foreign materials, such as dust and bacteria, from entering the eye.

The keratin 12 protein partners with another keratin protein, keratin 3, to form molecules known as intermediate filaments. These filaments assemble into strong networks that provide strength and resilience to the corneal epithelium.

### Health Conditions Related to Genetic Changes

#### Meesmann corneal dystrophy

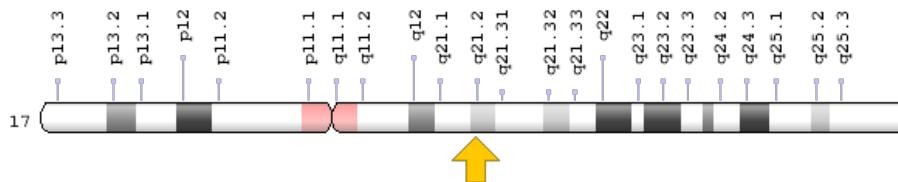
At least 20 mutations in the *KRT12* gene have been found to cause Meesmann corneal dystrophy, an eye disease characterized by the formation of tiny cysts in the corneal epithelium.

Almost all of the *KRT12* gene mutations associated with Meesmann corneal dystrophy change single protein building blocks (amino acids) in the keratin 12 protein. These changes occur in regions of the protein that are critical for the formation and stability of intermediate filaments. The altered keratin 12 protein interferes with the assembly of intermediate filaments, weakening the structural framework of the corneal epithelium. As a result, this outer layer of the cornea is abnormally fragile and develops the cysts that characterize Meesmann corneal dystrophy. The cysts likely contain clumps of abnormal keratin proteins and other cellular debris. When the cysts break open (rupture), they cause eye irritation, increased sensitivity to light (photophobia), and related symptoms.

## Chromosomal Location

Cytogenetic Location: 17q21.2, which is the long (q) arm of chromosome 17 at position 21.2

Molecular Location: base pairs 40,861,178 to 40,867,210 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- CK-12
- cytokeratin-12
- K1C12\_HUMAN
- K12
- keratin-12
- keratin 12, type I
- keratin, type I cytoskeletal 12

## Additional Information & Resources

### Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Intermediate Filaments  
<https://www.ncbi.nlm.nih.gov/books/NBK21560/>
- National Eye Institute: Facts About the Cornea and Corneal Disease  
<https://nei.nih.gov/health/cornealdisease/>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28KRT12%5BTIAB%5D%29+OR+%28keratin+12%5BTIAB%5D%29%29+OR+%28%28CK-12%5BTIAB%5D%29+OR+%28cytokeratin+12%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

## OMIM

- KERATIN 12, TYPE I  
<http://omim.org/entry/601687>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=KRT12%5Bgene%5D>
- HGNC Gene Family: Keratins, type I  
<http://www.genenames.org/cgi-bin/genefamilies/set/608>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=6414](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6414)
- Human Intermediate Filament Database  
[http://www.interfil.org/details.php?id=NM\\_000223](http://www.interfil.org/details.php?id=NM_000223)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/3859>
- UniProt  
<http://www.uniprot.org/uniprot/Q99456>

## **Sources for This Summary**

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<https://ghr.nlm.nih.gov/gene/KRT12>

Reviewed: August 2012

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications  
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National Institutes of Health  
Department of Health & Human Services